Case Study

PRECISION MEDICINE

Professor Philip Beales



- ⁽¹⁾ Mann, S. P., Savulescu, J. & Sahakian, B. J. Facilitating the ethical use of health data for the benefit of society: Electronic health records, consent and the duty of easy rescue. Philos. Trans. R. Soc. Math. Phys. Eng. Sci. 374, (2016).
- ^[2] Balthazar, P., Harri, P., Prater, A. & Safdar, N. M. Protecting Your Patients' Interests in the Era of Big Data, Artificial Inteligence, and Predictive Analytics. J. Am. Coll. Radiol. JACR 15, 580–586 (2018).
- ^[3] Floridi, L. et al. Key Ethical Challenges in the European Medical Information Framework. Minds Mach. 1–17 (2018) doi:10.1007/s11023-018-9467-4.
- ^[4] Voigt, K. Social Justice, Equality and Primary Care: (How) Can 'Big Data' Help? Philos. Technol. 32, 57–68 (2019).
- ^[5] Holzinger, A., Haibe-Kains, B. & Jurisica, I. Why imaging data alone is not enough: Albased integration of imaging, omics, and clinical data. Eur. J. Nucl. Med. Mol. Imaging (2019) doi: 10.1007/s00259-019-04382-9.
- ⁽⁶⁾ Kleinpeter, E. Four Ethical Issues of "E-Health". IRBM 38, 245–249 (2017).
- [7] Chiauzzi, E. & Newell, A. Mental health apps in psychiatric treatment:A patient perspective on real world technology usage. J. Med. Internet Res. 21, (2019).

Precision medicine encompasses predictive, preventive, personalised and participatory medicine (also termed P4 medicine)¹. It is moving from the traditional one-size-fits-all form of medicine to more preventative, personalised, data-driven disease management model that achieves improved patient outcomes and more cost-efficiencies. Precision medicine, as defined by the National Institute of Health (NIH), is an emerging approach for disease treatment and prevention that considers individual variability in genes, environment, and lifestyle². The promise is that precision medicine will more accurately predict which treatment and prevention strategies will work best for a particular patient.

There are, however, significant challenges to the widespread adoption of precision medicine that include a deluge of medical data, a paucity of trained specialists, and the enormous costs of drug development³. Take, for example, the 30% rise in the number of CT scans ordered in the UK between 2013 and 2016⁴ during which period the number of radiologists only increased by 3% annually⁵. Many studies demonstrate that as radiologists are compelled to work faster their interpretation error rate rises⁶.

However, AI can address this by leveraging deep learning approaches to overcome the obstacles inherent in large data sets and unstructured data. In clinical settings, AI can assist clinicians to work more efficiently and make more accurate diagnoses improving the productivity of healthcare workers.

In the broader setting, AI is helping industry to accelerate drug development, cut costs and gain faster approvals while reducing errors. According to a recent research report⁷, achieving the full potential of precision medicine will be impossible without applying AI and machine learning. Specifically, leveraging advanced machine learning and deep learning technology can outperform clinicians and researchers in rapidly analysing large datasets and integrating exponentially growing amounts of data from a wide variety of novel -omics sources (e.g. genomics, transcriptomics, proteomics, metabolomics, microbiomics) into clinically actionable insights

^[8] Krutzinna, J., Taddeo, M. & Floridi, L. Enabling Posthumous Medical Data Donation: An Appeal for the Ethical Utilisation of Personal Health Data. Sci. Eng. Ethics (2018) doi:10.1007/s11948-018-0067-8.

⁽⁹⁾ Shaw, D. M., Gross, J. V. & Erren, T. C. Data donation after death: A proposal to prevent the waste of medical research data. EMBO Rep. 17, 14–17 (2016).

^[10] Sterckx, S., Rakic, V., Cockbain, J. & Borry, P. "You hoped we would sleep walk into accepting the collection of our data": controversies surrounding the UK care.data scheme and their wider relevance for biomedical research. Med. Health Care Philos. 19, 177–190 (2016).. for personalised care plans and more effective population health management.

One challenge is how best to integrate precision medicine data into electronic health records (EHR). OSF HealthCare, an integrated healthcare network, has integrated CancerlQ's genetic cancer risk assessment program with Epic's EHR platform as part of a systemwide population health initiative to reduce cancer disparities and deaths⁸. In the US, individuals can now leverage Medfusion's national health data network to consolidate their health records and help drive discovery through private, secure data sharing using LunaPBC part of LunaDNA, the first community-owned genomic and health data platform – that enables members to access their EHRs⁹.

Al and machine learning are already transforming precision medicine delivery by driving computational phenotyping tools (<u>www.mendelian.co</u>) and new methods for target drug discovery (e.g. BenevolentAI). In fact there are now 100s of start-up companies using AI in drug discovery,¹⁰ demonstrating the power of this emerging sector for driving innovation in healthcare.

In clinical trials, fewer than a third of all phase II compounds make it to phase III, and one-third of phase III trials fail because the trial lacks enough patients or the right kinds of patients. AI can potentially boost the success rate of clinical trials by identifying and characterising patient subpopulations best suited for specific drugs and by efficiently measuring biomarkers that reflect the effectiveness of the drug being tested. Nevertheless, we are still at the proof-of-concept stage but feasibility pilot studies are demonstrating the high potential of numerous AI techniques for improving the performance of clinical trials.

Data-driven health will also likely impact patients directly by providing access to their own personal data, improving compliance with treatments and real time monitoring for adverse events making participation in P4 medicine a reality.